CYTOGENETICS OF DOMESTIC ANIMALS

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The chromosomal studies on domestic animals were initiated at the end of the last century; up to the decade of 1940 several experiments were carried out with the objective of establishing the chromosome number of various species. However, such studies were done on bone marrow, through a direct method without hypotonization, difficulting both the counting and morphological analysis of chromosomes.

The pre-treatment of cultivated cells with colchicine and hypotonic solution, carried out by Tjio and Levan (1956), initiated a new phase in cytogenetics, opening up new perspectives to the study of chromosomes, since it allowed their dispersion throught the cells with the apparent maintenance of shape and size. This phase culminated with the paper of Lejeune (1959) who, for the first time, related the karyotype with the phenotype by showing that, in man, the chromosomal abnormality kmown as trisomy 21 was related to the Down's Syndrome. At the same time, Jacobs and Strong (1959) showed that in the Klinefelter's Syndrome the carriers had three sex chromosomes and Ford et al. (1959) described the absense of one sex chromosome in individuals with the Turner's Syndrome.

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After these discoveries, a large number of congenital abnormalities were related with the description of new chromosomal aberrations, contributing significantly to the knowledge of those pathology's othiology in man.

The temporary culture of lymphocites from peripheral blood, after stimulation by phytohemagglutinin, discovered by Nowell (1960), being a simple method made the study of karyotypes of mammals even easier. From there on, the karyotypes of various species were established.

In domestic animals, after the discovery of the 1/29 robertsonian translocation in a herd of Red and White Swedish cattle (Gustavsson and Rockborn, 1964) and the relationship of this translocation with low fertility in that herd (Gustavsson, 1969), various cytogenetic studies were carried out in populations, mainly of bovine and sheep, with the objective of verifying if the chromosomal aberrations could have any interference on fertility and, as a consequence, to bring about economic losses.

However, the technique of conventional coloration by orcein did not possibilitate the correct pairing of the homologous chromosomes and their study becomes difficult, mainly in some species such as Bos taurus and Bos indicus where almost all chromosomes are acrocentric and their size decrease linearly, because their separation into groups is not possible. The introduction of the chromosomal banding techniques (Caspersson et al., 1968), with the use of quinacrine mustard, provided the identification of chromosome pairs by fluorescent regions (Q-bands), which are characteristics for each pair. From there on,

other studies were carried out establishing new techniques for chromosomal identification and marking of specific regions. Of great importance were the G-bands (Drets and Shaw, 1971), similar to the Q-bands, but having the advantage of being permanents and observable through the use of the common microscopy. Nowadays, besides these techniques, the R-bands are utilized after incorporating the thymidine analog 5-bromodeoxyuridine (BUdR) according to Dutrillaux et al. (1973), as well as the C-bands of constitutive heterochromatin (Arrighi and Hsu, 1971).

With the utilization of these techniques mentioned above, several chromosomal aberrations associated or not to reproductive failures and/or congenital abnormalities, were identified (Tables I and II). The incidence of the 1/29 robertsonian translocation in cattle populations is summarized in table III. In Brazil, cytogenetic studies were carried out on three breeds of cattle: Canchim, a 5/8 Charolais + 3/8 Zebu derived breed (Tambasco, 1976 and TAmbasco and Ferrari, 1986); Ibag, a 5/8 Angus + 3/8 Zebu derived breed (Pinheiro, 1977); and Pitangueiras, a 5/8 Red Polled + 3/8 Zebu derived breed (Pinheiro and Lobo, 1984). Only the Pitangueiras cattle had the 1/29 robertsonian translocation. The aberrations found in Canchim cattle are presented in Table IV.

Nowadays, cytogenetics studies are carried out as a routine for dettecting and eliminating carriers of chromosomal aberrations in populations of domestic animals. Besides its application in more sophisticated studies of genetic mapping, cytogentics can be useful to the technique of embryo transfer,

which has been exploited with good perpectives in the near future. The major application appears to be the possibility of diagnosting chromosomal aberrations, in early stages of embryo development, that can be due to errors in gametogenesis, external factors (e.g., superovulation) and transmitted aberrations; moreover, it would become possible to choose the sex of an embryo to be transferred and to eliminate the risk of producing freemartins in the case of twins. Therefore, cytogenetics showed be considered as an addicional tool in studies of reproduction and animal breeding.

TABLE I - SEX CHROMOSSOME ANEUPLOIDY (PURE, MOSAIC AND CHIMERIC) AND MAIN PHENOTYPIC EFFECTS IN THE GENERAL POPULATION OF DOMESTIC ANIMALS

SEX CHROMOSOMES	SPECIES	MAIN PHENOTYPIC EFFECTS	REFERENCE
XO	pig	intersexuality, ovarian hypoplasia	Nes 1960
	horse	ovarian hypoplasia	CHANDLEY et al. 1975, HUGHES and TROMMERSHAUSEN-SMITH 1977
	cat	(died before puberty)	NORBY et al. 1974
XO/XX	horse	ovarian hypoplasia	CHANDLEY et al. 1975, HUGHES and TROMMERSHAUSEN-SMITH 1977
XO/XX/XY	pig	intersexuality	LODJA 1969
XXX	cattle	no effects	RIECK et al. 1970
		ovarian hypoplasia	NORBERG et al. 1976
	horse	infertility	CHANDLEY et al. 1975
XXY	cattle	testicular hypoplasia	RIECK 1970
	sheep	testicular hypoplasia	BRUERE et al. 1969
	pig	testicular hypoplasia	BREEUWSMA 1968
	dog	testicular hypoplasia	CLOUGH et al. 1970
	cat	testicular hypoplasia	review see BENIRSCHKE 1972
XXY/XY	cattle	testicular hypoplasia	DOUTERS and VANDEPLASSCHE 1973
XXY/XX	cattle	intersexuality	DAIN and BRUDOE 1970
	pig	intersexuality	TOYAMA 1974
	horse	intersexuality	DOUTERS et al. 1972
	cat	testicular hypoplasia	review see BENIRSCHKE 1972
XXY/XX/XY	cattle	testicular hypoplasia	RIECK et al. 1969
XXY/XY/XO	cattle	testicular hypoplasia	LODJA and HAVRANKOVA 1975
XXY/XY/XX/XO	horse	cryptorchidism	BASRUR et al. 1969
XXXY	horse	intersexuality	GLUNOVSCHI et al. 1970
XXXY/XXY	pig	(no information)	HARVEY 1969
XYY/XY	cattle	no effects	DOBRYANOV and KONSTANTINOV 1970

GUSTAVSSON, 1980

TABLE II - CENTRIC FUSION TRANSLOCATIONS DESCRIBED IN CATTLE

DESIGNATION	BREED	COUNTRY OF DISTRIBUTION	REFERENCE
1/29	several	Several	review see GUSTAVSSON 1979a
mosaic 13/21/normal	Holstein-Friesian	Hungary	KOVCS et al. 1973
11-12/15-16 or 13/21	Simmental	New Zealand and Great Britain	BRUERE et CHAPMAN 1973, HARVEY and LOGUE 1975
7-11/20-25	Aquitaine Blond x Limousin	France	DARRE et al. 1974
5-6/15-16	Dexter	USA	ELDRIDRE 1974
2/4	British Friesian	Great Britain	POLLOCK and BOWMAN 1974
07/29	Guernsey	Canada	BONGSO and DASRUR 1976
1/25	Simmental	West-Germany	STRANZINGER and FORSTER 1976
3/4	Limousin	France	POPESCU 1977
8/?	Brown Swiss	Switzerland	TSCHUDI et al. 1977

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TABLE III - INCIDENCE OF 1/29 TRANSLOCATION IN DIFFERENT BREEDS OF CATTLE.

BREED	COUNTRY	NUMBER OF ANIMALS	TRANSLOCATION CARRIERS 1/29		REFERENCE
			NUMBER	%	
ABONDANCE	FRANCE	11	0	0	POPESCU, 1980
ARMORICAINE	FRANCE	10	0	0	POPESCU, 1980
BADOU	FRANCE	85	3	3.5	POPESCU, 1980
BLONDE D'AQUITAINE	FRANCE	228	47	20.6	GUEINNIC et al., 1974
BLONDE D'AQUITAINE	FRANCE	46	6	13.0	POPESCU, 1980
BROWN SWISS AMERICAN	USA	224	3	1.3	ELDRIDGE & BLASAK, 1975
BROW SWISS	SWITZERLAND	430	1	0.2	TSCHUDI et al., 1977
BRUNE DES ALPES	FRANCE	7	0	0	POPESCU, 1980
CANCHIM	BRAZIL	626	0	0	TAMBASCO, 1980
CHAROLAIS	FRANCE	359	12	3.3	POPESCU, 1980
CHAROLAIS	BRAZIL	31	2	6.5	MORAES & MATTEVI, 1980
CHAROLAIS	GREAT BRITAIN	185	1	0.5	HARVEY, 1976
CROLE	GUADALOUPE FR. ANTILLES	49	1	2.0	POPESCU et al., 1987
EGYPTIAN-BALADI	FRANCE	12	0	0	POPESCU, 1980
FFPN	FRANCE	778	0	0	POPESCU, 1980
FRIESIAN	ITALY	244	1	0.4	BALLI, 1984
FRIESIAN-HOLSTEIN FRIESIAN	DUTCH	300	0	0	DOGMA et al., 1980

(continuação)

TABLE II) - INCIDENCE OF 1/29 TRANSLOCATION IN DEFFERENT BREEDS OF CATTLE.

BREED	COUNTRY	NUMBER OF ANIMALS	TRANSLOCATION CARRIERS 1/29		REFERENCE
			NUMBER	%	
HEREFORD	AUSTRALIA	600	0	0	HALNAN, 1976
HEREFORD	FRANCE	16	0	0	POPESCU, 1980
HEREFORD	BRAZIL	32	0	0	MORAES & MATTEVI, 1980
HOLSTEIN	FRANCE	399	0	0	POPESCU, 1980
HOLSTEIN-FRIESIAN (SWEDISH)	SWEDEN	101	0	0	GUSTAVSSON, 1969
HOLSTEIN-FRIESIAN	USA	740	0	0	FECHHEIMER, 1973
HOLSTEIN-FRIESIAN	GREAT BRITAIN	330	0	0	POLLOCK, 1974
HOLSTEIN-FRIESIAN	AUSTRALIA	174	0	0	HALNAN, 1976
HOLSTEIN-FRIESIAN	GREAT BRITAIN	586	0	0	HARVEY, 1976
HUNGARIAN GREY	HUNGARY	106	4	3.8	KOVACS & PAPP, 1977
LIMOUSIN	FRANCE	231	13	5.6	QUEINNEC et all, 1974
LIMOUSIN	FRANCE	133	10	7.5	POPESCU, 1980
MAINE-ANJOU	FRANCE	70	0	0	POPESCU 1987
MARCHIGIANA	BRAZIL	8	4	50.0	CARRARA et al., 1978
MONTBLIARD	FRANCE	345	8	2.2	POPESCU, 1980
MONTBLIARD	FRANCE	93	1	1.1	CRIOU, 1981
N'DAMA	FRANCE	26	0	0	POPESCU, 1980
NORMANDY	FRANCE	249	0	0	POPESCU, 1980
NORMANDY	FRANCE	312	0	0	POPESCU, 1980

(continuação)

TABLE 111 - INCIDENCE OF 1/29 TRANSLOCATION IN DIFFERENT BREEDS OF CATTLE.

BREED	COUNTRY	NUMBER OF ANIMALS	TRANSLOCATION CARRIERS 1/29		REFERENCE
			NUMBER	%	
NORMANDY	BRAZIL	20	0	0	MORAES & MATTEVI, 1980
NORWEGIAN RED	NORWAY	430	18	4.2	AMUND, 1967
PIED NOIRE	FRANCE	129	0	0	CRIBIU, 1981
PIEMONTESE	FRANCE	7	0	0	POPESCU, 1980
PITANGUEIRAS	BRAZIL	545	104	19.0	PINHEIRO & LOBO, 1984
ROMAGNOLA	ITALY	9	2	22.2	SUCCI et al., 1976
ROMAGNOLA	ITALY	122	39	32.0	MOLTENI et al., 1977
SIMMENTAL (GERMAN) FLECKVIEH	WEST GERMANY	100	0	0	HOHN & HERZOG, 1976
SIMMENTAL	GREAT BRITAIN	113	3	2.7	HARVEY, 1976
SIMMENTAL	SWITZERLAND	454	21	9.2	TSCHUDI et al., 1977
SIMMENTAL (FRANCE) PIE ROUGE	FRANCE	34	0	0	POPESCU, 1980
SIMMENTAL	BRAZIL	5	0	0	MORAES & MATTEVI, 1980
SYCHEVKA	USSR	145	20	13.8	ZHIBACHEV et al., 1984
SWEDISH RED AND WHITE	SWEDEN	1173	164	14.3	GUSTAVSSON, 1967
SWEDISH RED AND WHITE	SWEDEN	944	120	12,4	GUSTAVSSON, 1971
TARENTAISE	FRANCE	10	0	0	POPESCU, 1980
VOSGIENNE	FRANCE	81	1	1.2	POPESCU, 1980
ZEBU AND CROSSBRED	FRANCE	51	3	5.9	POPESCU, 1980

TABLE IV - CHROMOSOME ABERRATIONS IN CANCHIM CATTLE

(11 ANIMALS -1.76% - IN A HERD OF 626)

CHROMOSSOME ABERRATIONS	NUMBER OF ANIMALS	PHENOTYPE
60,XY / 61,XY + mar	1	Normal
60,XX - gap X	1	Normal
60,XY chromossome breaks	2	Normal
60,XY chromossome breaks	3	Andrological problems
60,XX/ 60,XY	2	Freemartin
60,XX/ 61,XXY	1	Freemartin
60,XY + t 17/21-22	1	Normal

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